

## SESSION IV

*Chairman:* PROFESSOR A. I. DARLING

### Van Buchem disease

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#### Summary

Van Buchem disease is a hereditary sclerosing dysplasia of bone. Both dominant and autosomal recessive modes of transmission have been described. The dominant form tends to be a benign disorder and symptoms are usually confined to those associated with the enlargement of the jaw. The recessive forms tend to have a greater morbidity and symptoms arise from pressure on cranial nerves by hyperostotic bone at the base of the skull. Patients of the dominant families have often had a torus palatinus.

No haematological changes are found. The alkaline phosphatase may be raised—even if the total level is not elevated, the bone fraction may be increased.

The radiological appearances are regarded as characteristic. The jaw is enlarged and thickened to an extent not seen in other bone dysplasias such as osteopetrosis. The cortices of the diaphyses are thickened and the medullary cavities are encroached upon but not obliterated. Abnormal modelling of the bone ends is not found in van Buchem disease. In long bones the distribution is predominantly diaphyseal but the bone ends are also affected.

In 1955 van Buchem, Hadders and Ubbens of Groningen in the Netherlands described a peculiar systemic disease of the skeleton in a twin brother and sister. They stated 'This peculiar systemic disease, notwithstanding accurate clinical observation over a number of years and, in one of the cases, extensive post-mortem examination, has not fitted into any classification of diseases of the skeleton so far described'. Some signs which also occur in other known bone diseases were present although, in these, many dissimilarities were found. Features

emphasized in the brother and sister were enlargement of the jaw, hyperostosis of the skull, deafness and visual changes. The ribs were thickened as were the clavicles; and hyperostosis of the long bones, predominantly of the diaphyses, was reported.

They designated the disease 'hyperostosis corticalis generalisata familiaris', but the eponymous title 'van Buchem disease' is still often applied. Spranger, Langer and Wiedemann (1974) have called the condition 'endosteal hyperostosis'. The writer believes that this term is not as embracing as it might be; it notes only one aspect of the condition.

The original authors gave a very clear differential diagnosis. In 1962 van Buchem and his colleagues added five more cases to their previous two and in 1971 van Buchem added a further eight cases. Cases have been described from as far afield as America, Canada and France.

The author's own interest in this disease was aroused when some radiographs were shown to him at a meeting in Portsmouth in 1973. A boy of 17 years had injured his right knee. No fracture was found but the presence of an underlying bony abnormality prompted a skeletal survey.

#### X-rays

*Skull* (Fig. 1). Gross thickening of the calvarium, particularly in the frontal and basi-occipital regions. The skull base and petrous bones were very thickened. The maxilla was very hyperostotic. Unfortunately the mandible was not included on the radiograph and the patient would not attend for re-examination. The frontal sinuses were just visible. An abnormality was also found in the cervical spine.

*Knees* (Fig. 2). A textural change was seen in the shafts and an abnormal pattern was shown in the



FIG. 1. van Buchem disease skull. General hyperostosis of the vault, base and maxilla. Obliteration of the frontal sinus is seen.

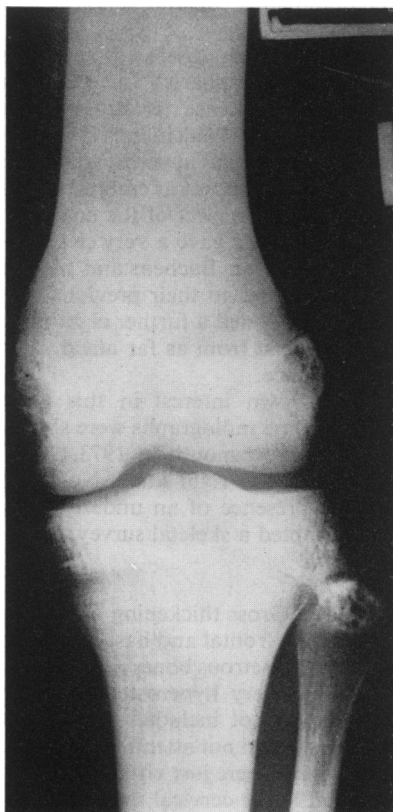


FIG. 2. van Buchem disease knee. Note that the hyperostosis extends to the ends of the bones.

metaphyses and epiphyses. The author particularly wishes to emphasize the epiphyseal changes.

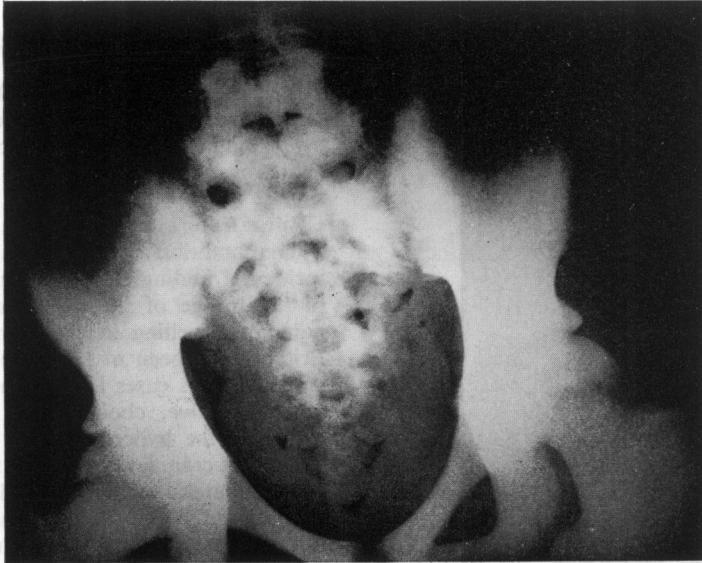
*Pelvis* (Fig. 3). A general amorphous density was seen. The upper ends of the femora were affected, including the epiphyses of the femoral heads.

*Hands* (Fig. 4). Sclerosis of the diaphyses of metacarpals and phalanges was shown. The sclerosis extended to the metacarpal bases and to the heads of proximal and middle phalanges. Textural changes were also seen in the carpal bones.

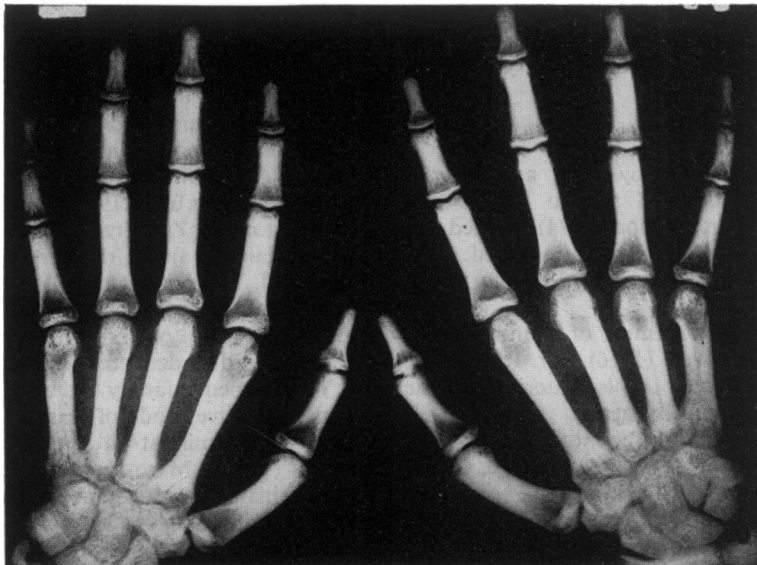
*Spine* (Fig. 5). The lower dorsal and lumbar vertebral bodies had a uniform outline but the middle thirds of the vertebral bodies were dense. The outer thirds were of normal density. The dense areas spread back into the posterior arches.

The patient was asymptomatic. Limited biochemical investigations showed a normal serum calcium and phosphate, but the alkaline phosphatase was raised to 23 KAu. Clinically he had some frontal bossing, a prominent jaw, a broad nose and widely separated eyes. His intelligence was described as average. He had been discharged from the army when a boy soldier because of the discovery of a bone disease.

The local radiologists had suggested a diagnosis of osteopetrosis (Albers-Schönberg disease) and had received support for this diagnosis from some distinguished colleagues. The author felt that a diagnosis of Albers-Schönberg disease could not be supported and after reference to the literature suggested a diagnosis of van Buchem disease; this diagnosis was confirmed by Professor van Buchem (1973).



**FIG. 3.** *van Buchem disease* pelvis. General amorphous density: note involvement of the femoral heads.



**FIG. 4.** *van Buchem disease* hands. Sclerosis primarily of the diaphyses extending to the proximal ends of the metacarpals and the distal ends of the proximal and middle phalanges. Diminution of the medullary canals is seen. Textural changes also noted in the carpal bones.

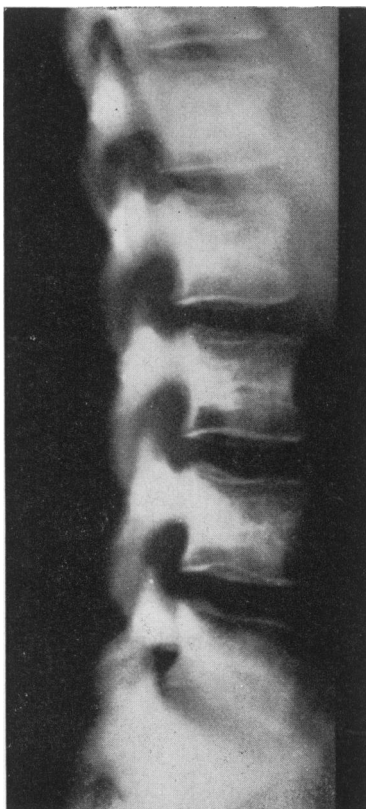


FIG. 5. van Buchem disease spine. Sclerosis of middle thirds of vertebral bodies extending back to the pedicles.

The case was published by Scott and Gautby (1974). The author believed this to be the first reported in Britain but Watson (1974) pointed out that a case had been previously described. This patient was reported by Dyson (1972) in the *British Journal of Oral Surgery*.

Dyson's patient was a member of a family of six, published later by Owen (1976). Dr Rex Owen kindly allowed me access to his material and the family had the following changes:

(1) Female aged 64 years, the mother of Dyson's patient—rectangular face, wide nasal bridge, square jaw. Hyperostosis of vault and base of skull, mandible, ribs and scapulae.

(2) Female aged 40 years, Dyson's patient—similar facial and skull findings to those of mother. Increased density and thickness of cortices of long bones with reduction of medullary canals.

(3) Male aged 19 years, the son of (2)—similar changes to those of mother.

(4) Male aged 16 years, the son of (2)—similar changes to those of mother.

(5) Male aged 14 years, the son of (2)—no lesion found.

(6) Female aged 10 years, the daughter of (2)—changes in skull, mandible, upper 7 ribs and clavicles.

(7) Male aged 7 years, the son of (2)—changes in skull, mandible and ribs.

### Clinical features

The condition may be benign and discovered as an incidental finding. Patients complain of thickening of the bridge of the nose, nasal obstruction and pain and swelling in the mandible. The cases in Britain have been of this type. However, many of van Buchem's cases had deafness, both conductive and perceptive, choked discs and facial paralysis owing to the hyperostosis in the skull base compromising cranial nerves. The facial paralysis tends to be unilateral in children and bilateral in adults.

It should be emphasized that these patients do not get prognathism and that the teeth are set normally. The sinuses tend to be underdeveloped and air spaces are later occluded by the lesion. Any similarity to acromegaly is clinical and not radiological.

It is noteworthy that no patient with van Buchem disease has suffered from an excessive tendency to fractures. This is an important differentiating point from osteopetrosis—a condition frequently complicated by pathological fractures.

### Genetic aspects

One family of eight patients described by van Buchem (1971) all lived in Urk in The Netherlands. The demographic history of the population of this former island is very interesting. In 1637, only 151 of the 300 inhabitants survived the plague and many of the marriages up to 1941 were endogamous. Then that part of the Zuyderzee in which Urk is situated was reclaimed and Urk was thus joined to the mainland. The present population may be regarded as descendants of about seventy-five marriages in the seventeenth century. Many other genetically determined disorders are found in this small area.

Van Buchem's family had the pattern of an autosomal recessive mode of transmission, whereas the trait was dominant in the family described by Owen (1976). Maroteaux *et al.* (1971) reported a dominant transmission in a French family. Worth and Wollin reported (1966) also a dominant transmission in nine members of a family of United Empire Loyalist stock who had settled in Prince Edward County, Canada, some 150–200 years previously. This family was closely knit but no proof of consanguinity was found.

Some of the patients in the families reported by

Worth and Wollin (1966) and by Maroteaux *et al.* (1971) had a torus palatinus. This is an osseous hypertrophy of the median raphe of the palatine fossa of the maxilla. It is said to occur in some 5% of the normal population (and in 60% of Esquimaux). It usually causes no disability although it may interfere with the fitting of dentures. The similar torus mandibularis has also been reported in this disease. These lesions have familial tendencies.

#### Laboratory investigations

No haematological changes are found but the serum alkaline phosphatase is usually raised. Van Buchem (1970) stated that the bone fraction is also present even when the total alkaline phosphatase is not increased. The 24-hr hydroxyproline excretion is normal.

#### Pathology

There is deposition of excessive but normal bone which is lamellar but with narrow Haversian canals. A normal fibre pattern is seen. Endosteal thickening causes consequent narrowing of the medulla. Van Buchem *et al.* (1962) reported that Professor Sissons had studied microradiographs of one patient and he reported that the bone showed a normal variation in the degree of mineralization of different Haversian systems. The fact that new bone is normal is probably the reason why these patients do not suffer fractures as may be seen in the abnormal bone of osteopetrosis.

#### Radiological findings

##### (a) Skull

Hyperostosis of the vault, base, maxilla and mandible (Fig. 6). A torus palatinus may be visible. No prognathism and normal sella turcica.

##### (b) Thorax

Cortical thickening of the clavicles and of the ribs—especially the upper (Fig. 7). The medial end of the clavicles may be widened.

##### (c) Long bones

Symmetrical cortical thickening and extra density with endosteal involvement and diminution of the medullary canal (Fig. 8). Van Buchem (1962) emphasized the diaphyseal involvement but stated that bone ends were not affected, so also did Fosmoe, Holm and Roscoe (1968). This statement was not supported by some of the illustrations of those authors. Other patients have shown clear evidence of epiphyseal involvement.

##### (d) Pelvis

In some patients the pelvis is uniformly dense (Fig. 3); in others it is unaffected or minimally affected. Owen (1976) has noted a small star-like area of sclerosis above the acetabula in some of his patients (Fig. 9).

##### (e) Spine

The spine is not affected in all patients. However, should the spine be affected, then the appearances may be characteristic. Sclerosis is seen in the middle third of vertebral bodies—‘the reverse sandwich vertebra’—the opposite to that seen in osteopetrosis. Sclerosis may extend to the posterior arches.

##### (f) Hands and feet

Characteristic appearances may be found in the hand. Cortical thickening, endosteal sclerosis—primarily diaphyseal may be seen but the lesion tends to spread to the bone ends—especially the distal ends of the phalanges and the proximal ends

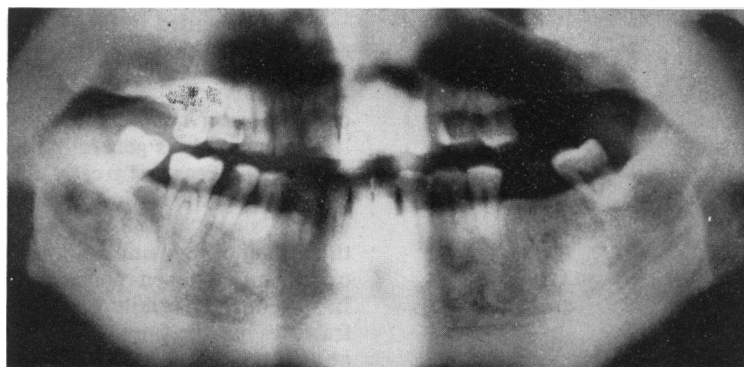


FIG. 6. van Buchem disease mandible showing generalized hyperostosis.

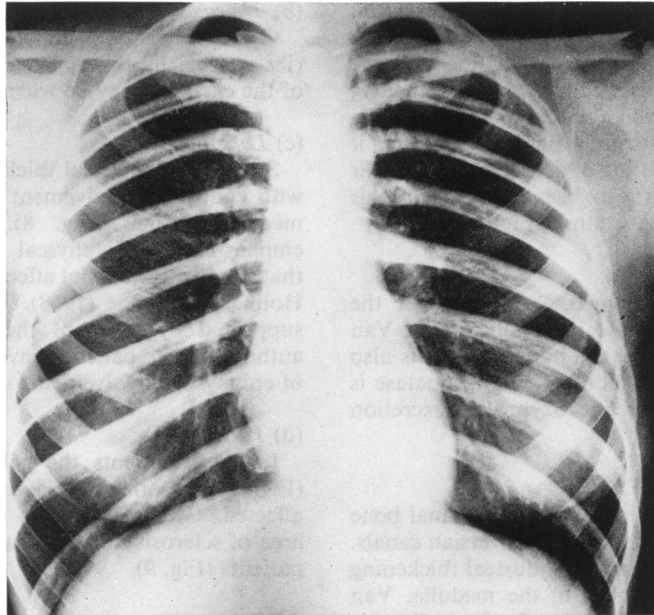


FIG. 7. van Buchem disease. Note cortical thickening in the upper ribs and in the clavicle.



FIG. 8. van Buchem disease long bones showing reduction of medullary canals.

of the metacarpals. Textural changes are also present in carpal bones.

#### **Differential diagnosis**

##### *Acromegaly*

The resemblance is clinical because of the jaw changes and not radiological. In van Buchem disease no prognathism or interference with the set of the teeth is seen and sinuses tend to be occluded rather than enlarged.

##### *Osteitis deformans (Paget's disease)*

This is included in many accounts but it has no real resemblance to van Buchem disease.

##### *Osteopetrosis (Albers-Schönberg disease)*

The mandible is generally little affected in osteopetrosis, whereas it is greatly affected in van Buchem disease. This distinction is only relative. Two noteworthy signs in the mandible in osteopetrosis, thickening of the lamina dura and of the mandibular canal, have not been reported in van Buchem disease. In osteopetrosis failure of bone modelling and fractures may be found. Moreover, the lesion begins at the bone ends and spreads to the diaphyses. The alkaline phosphatase is not raised in osteopetrosis.



FIG. 9. van Buchem disease pelvis. Normal except for stellate areas of hyperostosis above each acetabulum.

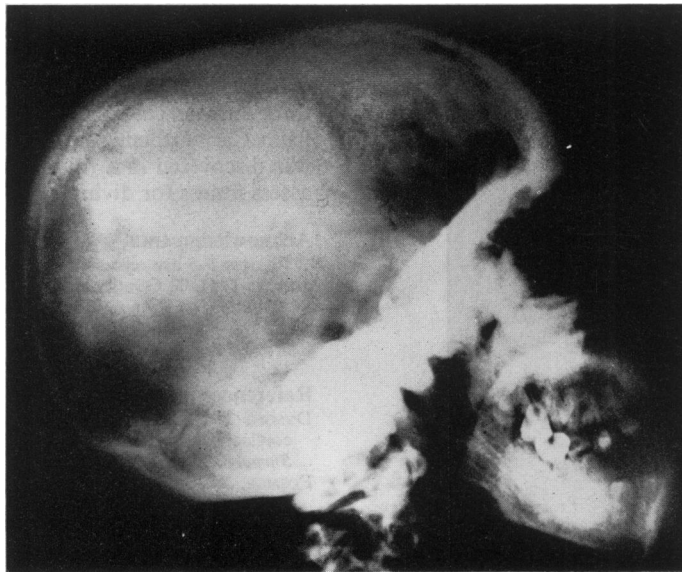


FIG. 10. Hyperostosis of the skull and mandible. Other bones were not affected except for their striated appearance (Fig. 11).



*Progressive diaphyseal dysplasia (Camurati-Engelmann disease)*

This may bear some resemblance. The diaphyses are mainly affected though the lesion may spread to the bone ends. The skull and mandible are usually little affected. These patients have a characteristic diminution in muscle mass.

*Osteopathia striata with generalized hyperostosis*

The diagnosis is made from the presence of a coarse striation in the long bones and pelvis.

*Osteopathia striata with skull thickening*

A condition of gross hyperostosis of the skull associated with osteopathia striata has been described. This is not, as is said, a combination of osteopetrosis and osteopathia striata but a distinct entity. The striated bones will enable the diagnosis to be made (Figs 10 and 11).

*Sclerosteosis*

These patients have thickening of the mandible and hyperostosis of tubular bones. However, syndactyly and absence of finger nails are found.



FIG. 11. Striations in the upper end of the tibia.

*Pachydermoperiostosis*

Little radiographic similarity is seen and these patients complain of joint changes.

*Myelosclerosis*

In this condition patchy sclerosis is seen but the skull is not affected and haematological changes are present.

*Halliday's hyperostosis*

This rare lesion was reported by Halliday (1950)—a single case of a 9-year-old girl with hyperostosis of the skull and facial bones. The mandible was less affected and the long bones were not hyperostotic.

**Discussion and conclusion**

Van Buchem disease is a rare hereditary disorder. Both dominant and recessive forms may be found. In the dominant type the symptoms are mild and mainly connected with thickening of the jaw. A torus palatinus may also be found in dominant types. The recessive form may be associated with much more morbidity owing to involvement of cranial nerves by the hyperostosis. Fractures and haematological changes are not found in van Buchem disease.

There are no specific laboratory findings. The total alkaline phosphatase may or may not be raised. Van Buchem says that the bone fraction of the alkaline phosphatase is always raised even when the total is not raised. Further confirmation of this observation is necessary.

The author believes that the X-ray findings are such that the condition should not be confused with other sclerosing lesions.

Since this paper was read the author has seen three further cases. Two were in a father and son from Carmarthen, south-west Wales, and the other was discovered in a routine medical examination to assess fitness for diving.

**Acknowledgments**

Figures 1–5 are reproduced by courtesy of Dr W. C. Scott and Dr T. H. T. Gautby and the Editor of the *British Journal of Radiology*; Figs 6 and 8 by courtesy of Dr Peter Jenkins, and Figs 7 and 9 by courtesy of Dr R. H. Owen and the Editor of the *British Journal of Radiology*.

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